



ALDH4A1 gene

aldehyde dehydrogenase 4 family member A1

Normal Function

The *ALDH4A1* gene provides instructions for producing the enzyme pyrroline-5-carboxylate dehydrogenase, which is found in tissues throughout the body. Within the cells of these tissues, this enzyme functions in energy-producing structures called mitochondria.

Pyrroline-5-carboxylate dehydrogenase starts the second step in the process that breaks down the protein building block (amino acid) proline. This step converts pyrroline-5-carboxylate, which is produced in the first step, to the amino acid glutamate. The conversion between proline and glutamate is important in maintaining a supply of the amino acids needed for protein production, and for energy transfer within the cell.

Health Conditions Related to Genetic Changes

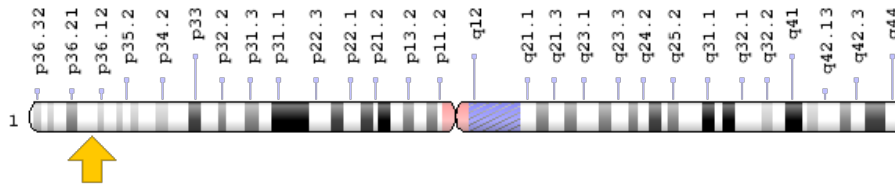
hyperprolinemia

Researchers have identified three mutations in the *ALDH4A1* gene that eliminate the function of the pyrroline-5-carboxylate dehydrogenase enzyme, causing hyperprolinemia type II. Two of these mutations add or delete a DNA building block (nucleotide), which introduces a premature stop signal that results in an abnormally shortened enzyme. The third mutation results in the substitution of the amino acid leucine for the amino acid serine at position 352 (written as Ser352Leu or S352L) in the pyrroline-5-carboxylate dehydrogenase enzyme. Nonfunctional pyrroline-5-carboxylate dehydrogenase leads to elevated levels of proline and a buildup of the intermediate breakdown product pyrroline-5-carboxylate, causing the signs and symptoms of hyperprolinemia type II.

Chromosomal Location

Cytogenetic Location: 1p36.13, which is the short (p) arm of chromosome 1 at position 36.13

Molecular Location: base pairs 18,871,430 to 18,902,799 on chromosome 1 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- AL4H1_HUMAN
- aldehyde dehydrogenase 4 family, member A1
- aldehyde dehydrogenase 4A1
- ALDH4
- mitochondrial delta-1-pyrroline 5-carboxylate dehydrogenase
- P5C dehydrogenase
- P5CD
- P5CDh
- P5CDhL
- P5CDhS

Additional Information & Resources

Educational Resources

- Sequence-Evolution-Function (2003): Proline Biosynthesis
<https://www.ncbi.nlm.nih.gov/books/NBK20266/#A486>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28ALDH4A1%5BTIAB%5D%29+OR+%28%28P5CD%5BTIAB%5D%29+OR+%28ALDH4%5BTIAB%5D%29+OR+%28P5CDh%5BTIAB%5D%29+OR+%28P5CDhS%5BTIAB%5D%29+OR+%28P5C+dehydrogenase%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- ALDEHYDE DEHYDROGENASE, FAMILY 4, SUBFAMILY A, MEMBER 1
<http://omim.org/entry/606811>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_ALDH4A1.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=ALDH4A1%5Bgene%5D>
- HGNC Gene Family: Aldehyde dehydrogenases
<http://www.genenames.org/cgi-bin/genefamilies/set/398>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=406
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/8659>
- UniProt
<http://www.uniprot.org/uniprot/P30038>

Sources for This Summary

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- Hu CA, Lin WW, Valle D. Cloning, characterization, and expression of cDNAs encoding human delta 1-pyrroline-5-carboxylate dehydrogenase. *J Biol Chem.* 1996 Apr 19;271(16):9795-800.
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<https://ghr.nlm.nih.gov/gene/ALDH4A1>

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